STUDIES OF SERUM TRANSAMINASES IN XERODERMA PIGMENTOSUM*

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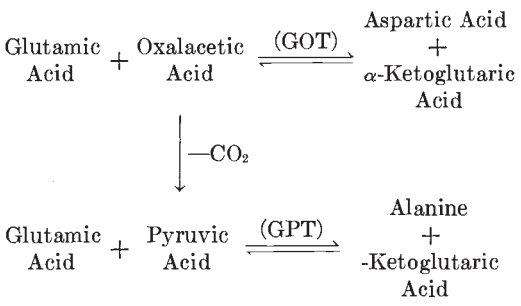
At present it is assumed that intracellular metabolism is essentially a collective chain of specific biochemical processes, each mediated by specific biologic catalysts, and that promotion of cell life depends upon the continuous activity of these agents (Dixon and Webb (1)). Further, it is postulated that enzyme contents of serum represent enzyme in transit from living healthy cells.

Alterations in enzyme activity in serum, therefore, reflect and depend upon physiologic alterations or actual destruction of selective tissues with consequent interference with intracellular metabolism and liberation of intracellular enzymes into this accessible channel. A rise in circulating content of an enzyme, therefore, may denote its escape as a result of damage of the primary source, the degree of elevation being related to both degree of damage and the relative concentration of enzyme in the primary source.

With this concept and with the availability of several sensitive tests, quantitative estimations of enzymatic activity in biologic fluids have been utilized clinically in diagnosing and assessing the prognosis of many diseases. Transaminases are enzymes which catalyze the reversible transfer of an alpha amino group from an amino acid to an alpha keto acid.



Serum glutamic oxalacetic transaminase (SGOT) and serum glutamic pyruvic transaminase (SGPT) are two of these important enzymes. They are present in considerable amounts in animal and human tissues; and in lesser amounts in the blood (2). The liver is the second organ after the heart in its concentration of the enzyme SGOT (3, 4). Liver damage (5, 6, 7, 8) and metastatic malignancy (9, 10) have been found to be associated with increase in SGOT. The values of the enzymatic activity decrease with clinical improvement and rise again during relapses (11). Pathologic alterations in transaminase activity can be produced by cardiac disease (12, 13, 14), muscular disease (15, 16, 17), shock or any acute hemolysis (4). SGPT in tissues has been found in the following descending order of concentration: liver, kidney, heart skeletal muscles, pancreas and serum (18).

The enzymes have been demonstrated in skin by Marsilii and Branzi (19). Only a few reports (20, 21) have been made on transaminases in skin diseases. Weber (1958, 1959) studied SGOT in serum and cantharidin blister fluid and in scales in thirty patients with skin diseases. Weber and Theisen (22) have also studied SGPT in twenty cases of dermatitis and nine sufferers from bullous diseases. De Morages, Perry and Fleischer (23), Siekert and Fleischer (15) and Moore, Birchall, Hoarack, and Bateson (24) studied SGOT in a number of cases of active dermatomyositis. Tickner, Mier and McCabe (25) studied SGOT and SGPT in a group of skin diseases (psoriasis, eczema, exfoliative dermatitis, atopic dermatitis, seborrheic dermatitis, lichen planus, pemphigoid, pemphigus vulgaris, systemic and discoid L.E., mycosis fungoides, dermatomyositis, Hodgkins disease, epithelioma, photosensitivity, dermatitis papulosa nigra, vitiligo, and pemphigus foliaceus).

In spite of the considerable work done in testing the activity of transaminases in various pathological conditions and the few reports on transaminases in certain skin diseases, no studies have been carried out on cases of xeroderma pigmentosum (x.d.p.). Having 33 cases of x.d.p. available for study we thought an investigation

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TABLE I

Serum glutamic oxalacetic transaminase in 33 cases x.d.p., 30 cases of their normal relatives and 10 control normal subjects

Case No	Normal Control	Normal Relatives			X.D.P.	
		Fathers	Mothers	Brothers & sisters	Mild cases	Severe cases
1	8.7	14.6	10.7	12.8	60.4	71.4
2	4.3	5.1	7.2	5.9	40.1	69.1
3	13.1	9.3	13.6	15.4	59.2	84.2
4	21.2	30.0	20.5	27.8	28.1	67.4
5	15.8	4.5	8.9	19.2	48.5	58.4
6	19.4	15.3	34.4	8.4	82.6	26.5
7	14.6	4.9	14.8	11.2	19.6	95.3
8	26.3	7.2	5.9	16.7	73.1	82.1
9	11.8	20.1	6.9	14.9	11.9	104.0
10	19.2	10.3	11.8	5.3	25.6	53.4
11					62.4	33.4
12					14.5	60.5
13					38.1	81.1
14					44.2	73.8
15					40.2	59.0
16					46.6	124.2
17						71.7
Range	4.3–26.3	4.5–30.0	5.9–34.4	5.3–27.8	11.9–82.6	26.5–124.2
Mean value	15.4	12.1	13.5	13.8	43.4	71.5

TABLE II

Serum glutamic pyruvic transaminase in 33 cases of x.d.p., 30 cases of their normal relatives and in 10 control normal subjects

Case No.	Normal Control	Normal Relatives			X.D.P.	
		Fathers	Mothers	Brothers & sisters	Mild cases	Severe cases
1	12.2	9.2	5.6	15.9	81.1	53.6
2	6.8	15.4	13.2	6.3	35.2	108.2
3	9.1	6.1	9.7	15.4	75.4	64.5
4	11.4	21.0	14.3	10.1	20.2	47.6
5	18.2	7.3	6.8	4.9	36.5	62.4
6	8.7	24.0	15.4	7.1	109.6	56.4
7	4.5	5.2	24.9	15.2	19.7	118.5
8	13.1	10.5	11.8	22.9	80.5	41.2
9	23.6	4.8	16.9	5.5	38.6	151.4
10	4.0	8.3	11.5	19.1	9.8	51.4
11					78.6	66.4
12					54.3	99.4
13					16.1	63.5
14					29.3	66.3
15					44.8	57.2
16					53.0	140.4
17						88.7
Range	4.0–23.6	4.8–21.0	5.6–24.9	4.9–22.9	9.8–109.6	41.2–151.4
Mean values	11.2	11.2	13.0	12.2	48.9	78.7

of enzymes of the skin in this disease and an attempt to correlate the findings with the clinical course would be of interest.

MATERIAL AND METHOD

The subjects for the study were 33 patients with xeroderma pigmentosum (x.d.p.) of ages ranging from 4–32 years (El-Hefnawi *et al* (26, 27, 28). Nineteen were males and 14 females. Of their parents and normal relatives, thirty patients were also studied. Ten normal subjects of both sexes were selected for the control group. These control subjects were carefully examined to make sure that they had had no disease past or present that might affect the estimations. Fasting venous blood samples were collected. All samples of blood were taken with a dry needle and syringe at 8 a.m. The blood was placed in a dry test tube, allowed to clot and the serum was separated by centrifugation.

The method used for the estimation of serum glutamic oxalacetic transaminase was that of Umbreit *et al* (29). Glutamic pyruvic transaminase was determined by the method of Reitman and Frankel (30).

A reference curve is constructed using a series of aqueous pyruvic acid solutions (as sodium pyruvate) ranging from 0–50 ug/ml. The results were expressed as colorimetric units. One unit is defined as the activity of 1 ml of serum which will form an amount of color complex in 20 minutes at 25° C (±2° C) equivalent to 1 ug/ml of pyruvic acid.

RESULTS

The results of SGOT in 10 normal controls ranged from 4.3 to 26.3 with a mean value of 15.4 units (this is in agreement with the normal range obtained for Egyptians in previous studies) (31, 32, 33). The results obtained for the normal relatives were found to be in the normal range. The results obtained ranged from 4.5 to 30.0, 5.9–34.4 and 5.3–27.8, with mean values of 12.1, 13.5 and 13.8 units respectively.

The values for SGOT in xeroderma pigmentosum patients were divided into two groups: The first group "A" consisted of 17 severe cases showing marked atrophy of skin, ulcerations, eye complications and malignancy. The results obtained for SGOT for this group ranged from 26.5 to 124.2 units with a mean value of 71.5 units. Only two cases had SGOT values near the higher level for normals or slightly above. Twelve cases (70.6%) showed SGOT results markedly higher than normal, with values ranging from 53.4–84.2 with a mean of 69.4 units; in three cases (17.6%) the values were extremely high 95.3, 104.0 and 124.2 units.

The second group "B" consisted of 16 mild cases demonstrating pigmentation, dryness, telangiectasia of exposed skin and some warty growths in addition to photophobia.

Six cases (37.5%) in this group gave SGOT values within normal range, five cases (31.25%) gave slightly raised values 40.1, 40.2, 44.2, 46.6 and 48.5, and the remaining five cases (31.25%) gave markedly raised values ranging from 59.2–82.6 with a mean of 67.5 units.

The values for SGPT in 10 normal controls ranged from 4.0 to 23.6 with a mean value of 11.2 units. This is in agreement with the normal range 5.0–21.0 with a mean of 10.7 obtained by Tickner *et al* (25) and 3.0–30.0 with a mean of 13.0 obtained by Velon and Santhanagopolan (34).

The results for the normal relatives were also found to be in the normal range, 4.8–21.0, 5.6–24.9 and 4.9–22.9 with mean values of 11.2, 13.0 and 12.2 respectively. In cases of group "A" of x.d.p. studied, the SGPT was found to be raised in 11 cases (64.7%) ranging from 41.2–66.4 with a mean of 57.3 and grossly raised in 6 cases (35.4%) ranging from 88.7–151.5 with an average of 117.8 units. In cases of group "B" of x.d.p., the SGPT was found to be within normal ranges in 4 cases (25.0%) with a mean value of 16.45, moderately increased in 4 cases (25.0%) with average of 29.3–38.6 and a mean of 34.9, markedly increased in 7 cases (43.7%) with a range of 44.8 to 81.1 with a mean of 66.8 units and grossly raised in one case (6.25%) with a value of 109.6 units.

DISCUSSION

The present study revealed that the SGOT and SGPT in the normal relatives of the 33 cases of xeroderma pigmentosum reported by us is within normal range. Our normal data are in agreement with the reports of previous workers in Egypt (31, 32, 33) and abroad (34). In the x.d.p. patients the usual routine liver function tests were done together with serum transaminases estimations. These liver function tests were cephalin cholesterol flocculation, bilirubin, alkaline phosphatase and thymol turbidity. The results were normal in all cases examined. Since the study of serum transaminases SGOT and SGPT is being widely used in the assessment

of hepatic function (Worblewski (35)), the finding of normal liver function tests in cases of x.d.p. studied indicates that the increase in the transaminases is not essentially of hepatic origin.

In the 33 cases of x.d.p. the SGOT was found to be within normal values in 18.2%, slightly increased in 21.2%, markedly increased in 51.5% and grossly increased in 9.1%. On the other hand the SGPT in the 33 cases of x.d.p. was found to be within normal in 12.1%, slightly increased in 12.1%, markedly increased in 54.5% and grossly increased in 21.3% of the cases. Tickner *et al* (25) studying SGOT and SGPT in a variety of skin diseases reported an increase of SGOT in 22% of the cases and an increase of SGPT in 11% of the cases. The findings of high SGOT and SGPT in the markedly photosensitive patients of x.d.p. to be in agreement with the findings of Tickner *et al* (25) in photosensitivity, not including x.d.p. Moreover the increase in SGOT and SGPT covered a greater percentage of the cases. Again we observed that the SGPT is more often affected than the SGOT. This observation is in agreement with that of Tickner *et al* (25) to the effect that SGPT is more often abnormal than SGOT in skin diseases. It is worthy of note that the extent of rise in SGOT and SGPT bears some relationship to the severity of the disease, early cases being the least affected.

SUMMARY

1. Serum glutamic oxalacetic transaminase (SGOT) and serum glutamic pyruvic transaminase (SGPT) were estimated in 33 cases of xeroderma pigmentosum (x.d.p.), 30 cases of their apparently normal relatives and 10 normal controls.

2. SGOT and SGPT values for the relatives were within normal.

3. SGOT was grossly increased in 9.1%, markedly increased in 51.5%, slightly increased in 21.2% and normal in 18.2% of the case of x.d.p.

4. SGPT was grossly increased in 21.3%, markedly increased in 54.5%, slightly increased in 12.1% and normal in 12.1% of the cases of x.d.p.

5. SGPT was more often affected than SGOT.

6. The extent of rise in SGOT and SGPT bears some relationship to the severity of the case.

7. Liver function tests yielded normal values in the cases of x.d.p., ruling out the possibility of hepatic origin for increment in serum transaminases.

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